Yusuke Okuno

List of Publications by Year
in descending order

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244	10,476	57631	37111
papers	citations	44 h-index	96 g-index

252	252	252	17004
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in SAM syndrome and palmoplantar keratoderma patients suggest genotype/phenotype correlations in DSG1 mutations. Journal of the European Academy of Dermatology and Venereology, 2022, 36, .	1.3	1
2	Minor PNH clones do not distinguish inherited bone marrow failure syndromes from immune-mediated aplastic anemia. Blood Advances, 2022, 6, 2517-2519.	2.5	4
3	EBV genome variations enhance clinicopathological features of nasopharyngeal carcinoma in a nonâ€endemic region. Cancer Science, 2022, , .	1.7	7
4	Elderlyâ€onset systemic Epsteinâ€“Barr virusâ€positive Tâ€cell lymphoma of childhood. Pathology International, 2022, 72, 376-378.	0.6	0
5	Direct reprogramming of adult adipose-derived regenerative cells toward cardiomyocytes using six transcriptional factors. IScience, 2022, 25, 104651.	1.9	3
6	Epsteinâ€“Barr virus tegument protein BGLF2 in exosomes released from virus-producing cells facilitates de novo infection. Cell Communication and Signaling, 2022, 20, .	2.7	9
7	Identification of Alpha Thalassemia, RNF 213 p.R4810K and PROC p.R189W among Children with Moyamoya Disease/Syndrome. Mediterranean Journal of Hematology and Infectious Diseases, 2022, 14, e2022057.	0.5	1
8	Detection of subclonal SETBP1 and JAK3 mutations in juvenile myelomonocytic leukemia using droplet digital PCR. Leukemia, 2021, 35, 259-263.	3.3	5
9	Phosphorylated proteome analysis of a novel germline ABL1 mutation causing an autosomal dominant syndrome with ventricular septal defect. International Journal of Cardiology, 2021, 326, 81-87.	0.8	2
10	Wholeâ€exome sequencing and host cell reactivation assay lead to a diagnosis of xeroderma pigmentosum group D with mild ultraviolet radiation sensitivity. Journal of Dermatology, 2021, 48, 96-100.	0.6	1
11	Role of Epsteinâ€“Barr Virus C Promoter Deletion in Diffuse Large B Cell Lymphoma. Cancers, 2021, 13, 561.	1.7	9
12	Clinical diagnostic value of telomere length measurement in inherited bone marrow failure syndromes. Haematologica, 2021, 106, 2511-2515.	1.7	6
13	RNAseq analysis identifies involvement of EBNA2 in PD-L1 induction during Epstein-Barr virus infection of primary B cells. Virology, 2021, 557, 44-54.	1.1	18
14	A patient with very early onset FH-deficient renal cell carcinoma diagnosed at age seven. Familial Cancer, 2021, , 1.	0.9	2
15	Relationship between plasma rabbit antiâ€thymocyte globulin concentration and immunosuppressive therapy response in patients with severe aplastic anemia. European Journal of Haematology, 2021, 107, 255-264.	1.1	3
16	Integrated diagnosis based on transcriptome analysis in suspected pediatric sarcomas. Npj Genomic Medicine, 2021, 6, 49.	1.7	8
17	Deletion of Viral microRNAs in the Oncogenesis of Epsteinâ€“Barr Virus-Associated Lymphoma. Frontiers in Microbiology, 2021, 12, 667968.	1.5	12
18	Mathematical Modeling and Mutational Analysis Reveal Optimal Therapy to Prevent Malignant Transformation in Grade II IDH-Mutant Gliomas. Cancer Research, 2021, 81, 4861-4873.	0.4	7

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19	Successful treatment of a novel type I interferonopathy due to a de novo PSMB9 gene mutation with a Janus kinase inhibitor. Journal of Allergy and Clinical Immunology, 2021, 148, 639-644.	1.5	23
20	Echocardiography Monitoring of Pulmonary Hypertension after Pediatric Hematopoietic Stem Cell Transplantation: Pediatric Pulmonary Arterial Hypertension and Pulmonary Veno-Occlusive Disease after Hematopoietic Stem Cell Transplantation. Transplantation and Cellular Therapy, 2021, 27, 786.e1-786.e8.	0.6	2
21	Simple and robust methylation test for risk stratification of patients with juvenile myelomonocytic leukemia. Blood Advances, 2021, 5, 5507-5518.	2.5	4
22	International Consensus Definition of DNA Methylation Subgroups in Juvenile Myelomonocytic Leukemia. Clinical Cancer Research, 2021, 27, 158-168.	3.2	35
23	Report on effective treatment and genetic predisposition in two children with refractory probable catastrophic antiphospholipid syndrome. Thrombosis Research, 2021, 208, 117-120.	0.8	1
24	Autoinflammatory Keratinization Disease With Hepatitis and Autism Reveals Roles for JAK1 Kinase Hyperactivity in Autoinflammation. Frontiers in Immunology, 2021, 12, 737747.	2.2	11
25	Targetable driver mutations in multicentric reticulohistiocytosis. Haematologica, 2020, 105, e61-e64.	1.7	11
26	A founder variant in the South Asian population leads to a high prevalence ofFANCLFanconi anemia cases in India. Human Mutation, 2020, 41, 122-128.	1.1	10
27	Novel compound heterozygous MCOLN1 mutations identified in a Japanese girl with severe developmental delay and thin corpus callosum. Brain and Development, 2020, 42, 298-301.	0.6	1
28	Oncogenesis of CAEBV revealed: Intragenic deletions in the viral genome and leaky expression of lytic genes. Reviews in Medical Virology, 2020, 30, e2095.	3.9	24
29	Novel biallelic FA2H mutations in a Japanese boy with fatty acid hydroxylase-associated neurodegeneration. Brain and Development, 2020, 42, 217-221.	0.6	8
30	Reduced stratum corneum acylceramides in autosomal recessive congenital ichthyosis with a NIPAL4 mutation. Journal of Dermatological Science, 2020, 97, 50-56.	1.0	16
31	Comprehensive pathogen detection in sera of Kawasaki disease patients by high-throughput sequencing: a retrospective exploratory study. BMC Pediatrics, 2020, 20, 482.	0.7	4
32	Multi-Lineage BCR-ABL Expression in Philadelphia Chromosome-Positive Acute Lymphoblastic Leukemia Is Associated With Improved Prognosis but No Specific Molecular Features. Frontiers in Oncology, 2020, 10, 586567.	1.3	7
33	Novel COL4A1 mutations identified in infants with congenital hemolytic anemia in association with brain malformations. Human Genome Variation, 2020, 7, 42.	0.4	2
34	Two Aldehyde Clearance Systems Are Essential to Prevent Lethal Formaldehyde Accumulation in Mice and Humans. Molecular Cell, 2020, 80, 996-1012.e9.	4.5	92
35	Genetic analysis in patients with newly diagnosed glioblastomas treated with interferon-beta plus temozolomide in comparison with temozolomide alone. Journal of Neuro-Oncology, 2020, 148, 17-27.	1.4	5
36	A novel sensitive detection method for DNA methylation in circulating free DNA of pancreatic cancer. PLoS ONE, 2020, 15, e0233782.	1.1	21

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37	H3F3A mutant allele specific imbalance in an aggressive subtype of diffuse midline glioma, H3 K27M-mutant. Acta Neuropathologica Communications, 2020, 8, 8.	2.4	14
38	Deep phenotyping of ichthyosis follicularis with atrichia and photophobia syndrome associated with MBTPS2 mutations. Journal of Dermatology, 2020, 47, e87-e88.	0.6	1
39	Frequent FOXA1-Activating Mutations in Extramammary Pagetâ€™s Disease. Cancers, 2020, 12, 820.	1.7	15
40	Direct Evidence of Abortive Lytic Infection-Mediated Establishment of Epstein-Barr Virus Latency During B-Cell Infection. Frontiers in Microbiology, 2020, 11, 575255.	1.5	27
41	Digenic mutations in ALDH2 and ADH5 impair formaldehyde clearance and cause a multisystem disorder, AMeD syndrome. Science Advances, 2020, 6, .	4.7	39
42	SDR9C7 catalyzes critical dehydrogenation of acylceramides for skin barrier formation. Journal of Clinical Investigation, 2020, 130, 890-903.	3.9	54
43	Peptides containing the MXXCW motif inhibit oncogenic RET kinase activity with a novel mechanism of action. American Journal of Cancer Research, 2020, 10, 336-349.	1.4	0
44	Diagnostic Whole Exome Sequencing for 166 Patients with Inherited Bone Marrow Failure Syndrome. Blood, 2020, 136, 9-9.	0.6	1
45	Study of pathophysiology and molecular characterization of congenital anemia in India using targeted next-generation sequencing approach. International Journal of Hematology, 2019, 110, 618-626.	0.7	14
46	DOCK8 mutation diagnosed using whole-exome sequencing of the dried blood spot-derived DNA: a case report of an Iraqi girl diagnosed in Japan. BMC Medical Genetics, 2019, 20, 114.	2.1	3
47	Next Generation Sequencing-Based Transcriptome Predicts Bevacizumab Efficacy in Combination with Temozolomide in Glioblastoma. Molecules, 2019, 24, 3046.	1.7	5
48	A novel CUL4B splice site variant in a young male exhibiting less pronounced features. Human Genome Variation, 2019, 6, 43.	0.4	4
49	Metagenomic analysis using next-generation sequencing of pathogens in bronchoalveolar lavage fluid from pediatric patients with respiratory failure. Scientific Reports, 2019, 9, 12909.	1.6	34
50	Pathogenic Epigenetic Consequences of Genetic Alterations in IDH-Wild-Type Diffuse Astrocytic Gliomas. Cancer Research, 2019, 79, 4814-4827.	0.4	6
51	Defective Epsteinâ€“Barr virus in chronic active infection and haematological malignancy. Nature Microbiology, 2019, 4, 404-413.	5.9	152
52	Essential role ofPTPN11mutation in enhanced haematopoietic differentiation potential of induced pluripotent stem cells of juvenile myelomonocytic leukaemia. British Journal of Haematology, 2019, 187, 163-173.	1.2	14
53	Elucidation of the pathogenic mechanism and potential treatment strategy for a female patient with spastic paraplegia derived from a single-nucleotide deletion in PLP1. Journal of Human Genetics, 2019, 64, 665-671.	1.1	9
54	KLF1 mutation E325K induces cell cycle arrest in erythroid cells differentiated from congenital dyserythropoietic anemia patient-specific induced pluripotent stem cells. Experimental Hematology, 2019, 73, 25-37.e8.	0.2	17

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55	Pathogenic mutations identified by a multimodality approach in 117 Japanese Fanconi anemia patients. Haematologica, 2019, 104, 1962-1973.	1.7	22
56	A Syrian Refugee in Iraq Diagnosed as a Case of IL12RB1 Deficiency in Japan Using Dried Blood Spots. Frontiers in Immunology, 2019, 10, 58.	2.2	6
57	Aberrant Active cis-Regulatory Elements Associated with Downregulation of RET Finger Protein Overcome Chemoresistance in Glioblastoma. Cell Reports, 2019, 26, 2274-2281.e5.	2.9	8
58	Transcriptome analysis offers a comprehensive illustration of the genetic background of pediatric acute myeloid leukemia. Blood Advances, 2019, 3, 3157-3169.	2.5	51
59	Frequent intragenic microdeletions of elastin in familial supravalvular aortic stenosis. International Journal of Cardiology, 2019, 274, 290-295.	0.8	15
60	Modification of cellular and humoral immunity by somatically reverted T cells in X-linked lymphoproliferative syndrome type 1. Journal of Allergy and Clinical Immunology, 2019, 143, 421-424.e11.	1.5	8
61	Associations of complementation group, ALDH2 genotype, and clonal abnormalities with hematological outcome in Japanese patients with Fanconi anemia. Annals of Hematology, 2019, 98, 271-280.	0.8	19
62	Utility of Newborn Screening for Severe Combined Immunodeficiency and X-Linked Agammaglobulinemia Using TREC and KREC Assays. Blood, 2019, 134, 3604-3604.	0.6	3
63	Comprehensive Mutational Analysis of Juvenile Myelomonocytic Leukemia Using Whole-Genome Sequencing. Blood, 2019, 134, 2974-2974.	0.6	0
64	Genome-Wide Methylation Analysis Using the Digital Restriction Enzyme Analysis of Methylation for Stratification of Patients with Juvenile Myelomonocytic Leukemia. Blood, 2019, 134, 2973-2973.	0.6	0
65	Detection of Subclonal SETBP1 and JAK3 Mutations in Patients with Juvenile Myelomonocytic Leukemia Using Droplet Digital PCR. Blood, 2019, 134, 4213-4213.	0.6	0
66	An infant with generalized pustular psoriasis and geographic tongue had a heterozygous IL36RN mutation and IgG2 deficiency. Journal of Dermatological Science, 2018, 90, 216-218.	1.0	2
67	Comprehensive detection of pathogens in immunocompromised children with bloodstream infections by next-generation sequencing. Scientific Reports, 2018, 8, 3784.	1.6	45
68	Dysregulation of Epstein-Barr Virus Infection in Hypomorphic ZAP70 Mutation. Journal of Infectious Diseases, 2018, 218, 825-834.	1.9	22
69	Integrated molecular profiling of juvenile myelomonocytic leukemia. Blood, 2018, 131, 1576-1586.	0.6	78
70	Mild case of Haileyâ€“Hailey disease caused by a novel <scp>ATP</scp>2C1 mutation. Journal of Dermatology, 2018, 45, e207-e208.	0.6	4
71	Enhanced Expression of Anti-CD19 Chimeric Antigen Receptor in piggyBac Transposon-Engineered T Cells. Molecular Therapy - Methods and Clinical Development, 2018, 8, 131-140.	1.8	49
72	A novel IFIH1 mutation in the pincer domain underlies the clinical features of both Aicardi-GoutiÃ¨res and Singleton-Merten syndromes in a single patient. British Journal of Dermatology, 2018, 178, e111-e113.	1.4	13

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73	Biallelic mutations in SZT2 cause a discernible clinical entity with epilepsy, developmental delay, macrocephaly and a dysmorphic corpus callosum. Brain and Development, 2018, 40, 134-139.	0.6	22
74	Genetic heterogeneity of uncharacterized childhood autoimmune diseases with lymphoproliferation. Pediatric Blood and Cancer, 2018, 65, e26831.	0.8	18
75	Trichothiodystrophy, complementation group A complicated with squamous cell carcinoma. Journal of the European Academy of Dermatology and Venereology, 2018, 32, e75-e77.	1.3	3
76	Comprehensive genetic analysis of donor cell derived leukemia with KMT2A rearrangement. Pediatric Blood and Cancer, 2018, 65, e26823.	0.8	4
77	Regulation of Epstein-Barr Virus Life Cycle and Cell Proliferation by Histone H3K27 Methyltransferase EZH2 in Akata Cells. MSphere, 2018, 3, .	1.3	25
78	Reduction of stratum corneum ceramides in Neu-Laxova syndrome caused by phosphoglycerate dehydrogenase deficiency. Journal of Lipid Research, 2018, 59, 2413-2420.	2.0	14
79	Gain-of-function IKBKB mutation causes human combined immune deficiency. Journal of Experimental Medicine, 2018, 215, 2715-2724.	4.2	69
80	Sterol profiles are valuable biomarkers for phenotype expression of Conradi-HÃ¼nermann-Happle syndrome with EBP mutations. British Journal of Dermatology, 2018, 179, 1186-1188.	1.4	3
81	A patient with a GNAO1 mutation with decreased spontaneous movements, hypotonia, and dystonic features. Brain and Development, 2018, 40, 926-930.	0.6	12
82	Whole-exome analysis to detect congenital hemolytic anemia mimicking congenital dyserythropoietic anemia. International Journal of Hematology, 2018, 108, 306-311.	0.7	8
83	Integration Mapping of piggyBac-Mediated CD19 Chimeric Antigen Receptor T Cells Analyzed by Novel Tagmentation-Assisted PCR. EBioMedicine, 2018, 34, 18-26.	2.7	30
84	Identification of potential pathogenic viruses in patients with acute myocarditis using nextâ€generation sequencing. Journal of Medical Virology, 2018, 90, 1814-1821.	2.5	25
85	De Novo Mutations Activating Germline TP53 in an Inherited Bone-Marrow-Failure Syndrome. American Journal of Human Genetics, 2018, 103, 440-447.	2.6	33
86	DNA Methylation Subgroups in Juvenile Myelomonocytic Leukemia: An International Collaborative Analysis and Development of a Common Diagnostic Platform. Blood, 2018, 132, 3093-3093.	0.6	2
87	Combination of TREC Measurement and Next-Generation Sequencing in Newborn Screening for Severe Combined Immunodeficiency: A Pilot Program in Japan. Blood, 2018, 132, 3717-3717.	0.6	5
88	Dopamine and Serotonin Receptors Cooperatively Modulate Pacemaker Activity of Intestinal Cells of Cajal. Chinese Journal of Physiology, 2018, 61, 302-312.	0.4	3
89	Analysis of Genomic Predispositions to Sporadic Myeloid Neoplasms Mediated By DDX41 in Japan. Blood, 2018, 132, 4371-4371.	0.6	0
90	Characterization of Pathogenic Variants and Clinical Phenotypes in 117 Japanese Fanconi Anemia Patients. Blood, 2018, 132, 3860-3860.	0.6	0

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91	The Presence of Defective Epstein-Barr Virus (EBV) Infection in Patients with EBV-Associated Hematological Malignancy. Blood, 2018, 132, 1562-1562.	0.6	0
92	Clinical and Genetic Characteristics of Patients with Shwachman-Diamond Syndrome in Japan. Blood, 2018, 132, 3862-3862.	0.6	0
93	Germline IKAROS mutation associated with primary immunodeficiency that progressed to T-cell acute lymphoblastic leukemia. Leukemia, 2017, 31, 1221-1223.	3.3	56
94	Clinical utility of next-generation sequencing for inherited bone marrow failure syndromes. Genetics in Medicine, 2017, 19, 796-802.	1.1	66
95	A case of GATA2â€related myelodysplastic syndrome with unbalanced translocation der(1;7)(q10;p10). Pediatric Blood and Cancer, 2017, 64, e26419.	0.8	6
96	Exome sequencing identified RPS15A as a novel causative gene for Diamond-Blackfan anemia. Haematologica, 2017, 102, e93-e96.	1.7	30
97	Common Variable Immunodeficiency Caused by FANC Mutations. Journal of Clinical Immunology, 2017, 37, 434-444.	2.0	18
98	Efficacy of neutrophil non-muscle myosin heavy chain-IIA immunofluorescence analysis in determining the pathogenicity of MYH9 variants. Annals of Hematology, 2017, 96, 1065-1066.	0.8	1
99	A combination of low-dose systemic etretinate and topical calcipotriol/betamethasone dipropionate treatment for hyperkeratosis and itching in Olmsted syndrome associated with a TRPV3 mutation. Journal of Dermatological Science, 2017, 88, 144-146.	1.0	5
100	Recurrent MYB rearrangement in blastic plasmacytoid dendritic cell neoplasm. Leukemia, 2017, 31, 1629-1633.	3.3	40
101	Autosomal dominant familial generalized pustular psoriasis caused by a CARD14 mutation. British Journal of Dermatology, 2017, 177, e133-e135.	1.4	27
102	Dynamics of clonal evolution in myelodysplastic syndromes. Nature Genetics, 2017, 49, 204-212.	9.4	348
103	Abnormal hematopoiesis and autoimmunity in human subjects with germline IKZF1 mutations. Journal of Allergy and Clinical Immunology, 2017, 140, 223-231.	1.5	99
104	Comprehensive detection of viruses in pediatric patients with acute liver failure using next-generation sequencing. Journal of Clinical Virology, 2017, 96, 67-72.	1.6	22
105	Loss of DNA Damage Response in Neuroblastoma and Utility of a PARP Inhibitor. Journal of the National Cancer Institute, 2017, 109, .	3.0	43
106	Biallelic Mutations in KDSR Disrupt Ceramide Synthesis and Result in aÂSpectrum of Keratinization Disorders Associated with Thrombocytopenia. Journal of Investigative Dermatology, 2017, 137, 2344-2353.	0.3	53
107	Development of clinical paroxysmal nocturnal haemoglobinuria in children with aplastic anaemia. British Journal of Haematology, 2017, 178, 954-958.	1.2	14
108	Haploinsufficiency of TNFAIP3 (A20) by germline mutation is involved in autoimmune lymphoproliferative syndrome. Journal of Allergy and Clinical Immunology, 2017, 139, 1914-1922.	1.5	91

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109	Clinical utility of nextâ€generation sequencingâ€based minimal residual disease in paediatric Bâ€cell acute lymphoblastic leukaemia. British Journal of Haematology, 2017, 176, 248-257.	1.2	32
110	A newly revealed IL36RN mutation in sibling cases complements our IL36RN mutation statistics for generalized pustular psoriasis. Journal of Dermatological Science, 2017, 85, 58-60.	1.0	15
111	Diagnostic challenge of Diamondâ€“Blackfan anemia in mothers and children by whole-exome sequencing. International Journal of Hematology, 2017, 105, 515-520.	0.7	18
112	A case of lamellar ichthyosis due to a novel TGM1 mutation associated with Parkinson's disease. European Journal of Dermatology, 2017, 27, 438-439.	0.3	1
113	Congenital Ichthyosis and Recurrent Eczema Associated with a Novel ALOXE3 Mutation. Acta Dermato-Venereologica, 2017, 97, 532-533.	0.6	4
114	Striate Palmoplantar Keratoderma Showing Transgrediens in a Patient Harbouring Heterozygous Nonsense Mutations in Both DSG1 and SERPINB7. Acta Dermato-Venereologica, 2017, 97, 399-401.	0.6	6
115	Next-generation sequencing-guided identification of causative gene mutations. Japanese Journal of Thrombosis and Hemostasis, 2017, 28, 3-8.	0.1	0
116	A Cytokineâ€Based Diagnostic Program in Pediatric Aplastic Anemia and Hypocellular Refractory Cytopenia of Childhood. Pediatric Blood and Cancer, 2016, 63, 652-658.	0.8	4
117	Functional characterization of a novel GFI1B mutation causing congenital macrothrombocytopenia. Journal of Thrombosis and Haemostasis, 2016, 14, 1462-1469.	1.9	31
118	705. Enhancement of CAR Expression of piggyBac Transposon-Engineered T Cells by Stimulation with Viral Antigens. Molecular Therapy, 2016, 24, S278-S279.	3.7	0
119	Immunosuppressive therapy for patients with Down syndrome and idiopathic aplastic anemia. International Journal of Hematology, 2016, 104, 130-133.	0.7	4
120	Application of extensively targeted next-generation sequencing for the diagnosis of primary immunodeficiencies. Journal of Allergy and Clinical Immunology, 2016, 138, 303-305.e3.	1.5	9
121	The phenotype and clinical course of Japanese Fanconi Anaemia infants is influenced by patient, but not maternal ALDH2 genotype. British Journal of Haematology, 2016, 175, 457-461.	1.2	10
122	Wholeâ€exome sequencing reveals the spectrum of gene mutations and the clonal evolution patterns in paediatric acute myeloid leukaemia. British Journal of Haematology, 2016, 175, 476-489.	1.2	60
123	Correlation of rabbit antithymocyte globulin serum levels and clinical outcomes in children who received hematopoietic stem cell transplantation from an alternative donor. Pediatric Transplantation, 2016, 20, 105-113.	0.5	16
124	MEF2D-BCL9 Fusion Gene Is Associated With High-Risk Acute B-Cell Precursor Lymphoblastic Leukemia in Adolescents. Journal of Clinical Oncology, 2016, 34, 3451-3459.	0.8	98
125	JAK2, MPL, and CALR mutations in children with essential thrombocythemia. International Journal of Hematology, 2016, 104, 266-267.	0.7	10
126	Phosphatase and tensin homolog (PTEN) mutation can cause activated phosphatidylinositol 3-kinase Î´ syndromeâ€“like immunodeficiency. Journal of Allergy and Clinical Immunology, 2016, 138, 1672-1680.e10.	1.5	87

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127	Successful T-cell reconstitution after unrelated cord blood transplantation in a patient with complete DiGeorge syndrome. Journal of Allergy and Clinical Immunology, 2016, 138, 1471-1473.e4.	1.5	4
128	Identification of Viruses in Cases of Pediatric Acute Encephalitis and Encephalopathy Using Next-Generation Sequencing. Scientific Reports, 2016, 6, 33452.	1.6	73
129	Markedly High Plasma Thrombopoietin (TPO) Level is a Predictor of Poor Response to Immunosuppressive Therapy in Children With Acquired Severe Aplastic Anemia. Pediatric Blood and Cancer, 2016, 63, 659-664.	0.8	13
130	PIEZO1 gene mutation in a Japanese family with hereditary high phosphatidylcholine hemolytic anemia and hemochromatosis-induced diabetes mellitus. International Journal of Hematology, 2016, 104, 125-129.	0.7	25
131	Extrapulmonary tuberculosis mimicking Mendelian susceptibility to mycobacterial disease in a patient with signal transducer andÂactivator of transcription 1 (STAT1) gain-of-function mutation. Journal of Allergy and Clinical Immunology, 2016, 137, 619-622.e1.	1.5	14
132	Development of Paroxysmal Nocturnal Hemoglobinuria in Children with Aplastic Anemia. Blood, 2016, 128, 1499-1499.	0.6	2
133	Novel and recurrent mutations in Japanese patients with Darier's disease. Nagoya Journal of Medical Science, 2016, 78, 485-492.	0.6	4
134	Plakin Family Autoantibodies in Bronchiolitis Obliterans Following Hematopoietic Stem Cell Transplantation As Useful Biomarkers and the Target for Rituximab Therapy. Blood, 2016, 128, 3432-3432.	0.6	0
135	Genetic Landscape and Clonal Evolution Following 5-Aza Therapy in Patients with High-Risk Myelodysplastic Syndromes. Blood, 2016, 128, 4304-4304.	0.6	0
136	Comprehensive Genetic Analysis in Cases of Juvenile Myelomonocytic Leukemia for Prognostic Estimation. Blood, 2016, 128, 3159-3159.	0.6	2
137	the Impact of Clonal Dynamics on Prognosis and Outcome in Myelodysplastic Syndromes. Blood, 2016, 128, 4287-4287.	0.6	0
138	Whole-Genome Sequencing of Primary Central Nervous System Lymphoma and Diffuse Large B-Cell Lymphoma. Blood, 2016, 128, 4112-4112.	0.6	2
139	Profiling of somatic mutations in acute myeloid leukemia with FLT3-ITD at diagnosis and relapse. Blood, 2015, 126, 2491-2501.	0.6	180
140	Mutations in the Gene Encoding the E2 Conjugating Enzyme UBE2T Cause Fanconi Anemia. American Journal of Human Genetics, 2015, 96, 1001-1007.	2.6	100
141	Paroxysmal nocturnal hemoglobinuria and telomere length predicts response to immunosuppressive therapy in pediatric aplastic anemia. Haematologica, 2015, 100, 1546-1552.	1.7	63
142	BRCC3 mutations in myeloid neoplasms. Haematologica, 2015, 100, 1051-7.	1.7	20
143	Choreito Formula for BK Virusâ€“associated Hemorrhagic Cystitis after Allogeneic Hematopoietic Stem Cell Transplantation. Biology of Blood and Marrow Transplantation, 2015, 21, 319-325.	2.0	18
144	Loss of function mutations in RPL27 and RPS27 identified by wholeâ€exome sequencing in Diamondâ€Blackfan anaemia. British Journal of Haematology, 2015, 168, 854-864.	1.2	87

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145	X-Linked Agammaglobulinemia Associated with B-Precursor Acute Lymphoblastic Leukemia. Journal of Clinical Immunology, 2015, 35, 108-111.	2.0	20
146	A Female Patient with Incomplete Hemophagocytic Lymphohistiocytosis Caused by a Heterozygous XIAP Mutation Associated with Non-Random X-Chromosome Inactivation Skewed Towards the Wild-Type XIAP Allele. Journal of Clinical Immunology, 2015, 35, 244-248.	2.0	28
147	Integrated genetic and epigenetic analysis defines novel molecular subgroups in rhabdomyosarcoma. Nature Communications, 2015, 6, 7557.	5.8	149
148	Somatic Mutations and Clonal Hematopoiesis in Aplastic Anemia. New England Journal of Medicine, 2015, 373, 35-47.	13.9	508
149	Somatic Mosaicism for a NRAS Mutation Associates with Disparate Clinical Features in RAS-associated Leukoproliferative Disease: a Report of Two Cases. Journal of Clinical Immunology, 2015, 35, 454-458.	2.0	30
150	Clinical and genetic features of dyskeratosis congenita, cryptic dyskeratosis congenita, and Hoyeraal-Hreidarsson syndrome in Japan. International Journal of Hematology, 2015, 102, 544-552.	0.7	21
151	GATA2 and secondary mutations in familial myelodysplastic syndromes and pediatric myeloid malignancies. Haematologica, 2015, 100, e398-e401.	1.7	48
152	Late-Onset Combined Immunodeficiency with a Novel IL2RG Mutation and Probable Revertant Somatic Mosaicism. Journal of Clinical Immunology, 2015, 35, 610-614.	2.0	26
153	Fulminant adenovirus hepatitis after hematopoietic stem cell transplant: Retrospective real-time PCR analysis for adenovirus DNA in two cases. Journal of Infection and Chemotherapy, 2015, 21, 857-863.	0.8	10
154	Two Novel Distinct Subtypes of Myeloid Neoplasms Molecularly Associated with Histone H3K36 Methylations. Blood, 2015, 126, 2841-2841.	0.6	1
155	Genetic Predispositions to Myeloid Neoplasms Caused By Germline DDX41 Mutations. Blood, 2015, 126, 2843-2843.	0.6	7
156	Genetic Background of Idiopathic Bone Marrow Failure Syndromes in Children. Blood, 2015, 126, 3610-3610.	0.6	2
157	Serial Sequencing in Myelodysplastic Syndromes Reveals Dynamic Changes in Clonal Architecture and Allows for a New Prognostic Assessment of Mutations Detected in Cross-Sectional Testing. Blood, 2015, 126, 709-709.	0.6	2
158	Aberrant DNA Methylation Is Associated with a Poor Outcome in Juvenile Myelomonocytic Leukemia. PLoS ONE, 2015, 10, e0145394.	1.1	25
159	Abstract 482: Integrated genetic and epigenetic analysis defines novel molecular clusters in rhabdomyosarcoma. , 2015, , .		0
160	COL4A1 is a Novel Causative Gene Responsible for Congenital Hemolytic Anemia, Representing Characteristic Clinical Course in Infants. Blood, 2015, 126, 934-934.	0.6	0
161	Landscape of DNA Methylation and Genetic Profiles in 291 Patients with Myelodysplastic Syndromes. Blood, 2015, 126, 5205-5205.	0.6	0
162	Functional Characterization of a Novel GFI1B Mutation Causing Congenital Macrothrombocytopenia. Blood, 2015, 126, 75-75.	0.6	0

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